COMPLETE ATRIOVENTRICULAR BLOCK IN BECKER MUSCULAR DYSTROPHY: A CASE REPORT

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SUMMARY

Cardiac involvement in Becker Muscular Dystrophy, including dilated cardiomyopathy, mild to moderate degree mitral regurgitation, cardiac conduction system abnormalities and various arrhythymias, is one of the leading problems during the progression of the disease (1,2). But, complete atrioventricular block associated with Becker Muscular Dystrophy which necessitates permanent pacemaker implantation is a rare condition. We reported a patient with Becker Muscular Dystrophy which complicated complete atrioventricular block and dilated cardiomyopathy. ***Arch Turk Soc Cardiol 2003;31:121-124***

Key Words: *Becker Muscular Dystrophy, Cardiomyopathy, Complete Atrioventricular Block.*

ÖZET

Becker Musküler Distrofisinde Tam Atrioventrikuler Blok: Olgu Sunumu

Becker Musküler Distrofi'de; dilate kardiyomiyopati, hafif-orta derecede mitral yetersizliği, kardiyak ritim ve ileti bozukluklarını içeren kardiyak tutulum, hastalığın seyri sırasında en önde gelen problemlerdendir(1,2). Ancak, Becker Muskuler Distrofi ile ilişkili, kalıcı kalp pili takılmasını gerektiren atrioventriküler tam blok oldukça nadir bir komplikasyondur. Bu yazıda, daha önceden Becker Muskuler Distrofi tanısıyla izlenmekte olan bir hastada saptanan dilate kardiyomiyopati ve atrioventriküler tam blok olgusu tartışılmaktadır. ***Türk Kardiyol Dern Arş 2003;31:121-124***

Anahtar Kelimeler: *Becker Muskuler Distrofi, Kardiyomiyopati, Atrioventriküler Tam Blok*

Becker muscular dystrophy (BMD) is a hereditary X-linked recessive disease. Mutations are located in the Xp21.2. There is frame deletion of the dystrophin gene. Clinically, it may be manifested between 5-40 years old or even later[1]. Initial symptom is commonly proximal muscle weakness especially, lower extremity muscles involved. Clinical or sub-clinical cardiac involvement may occur during the progression of the disease. Dilated cardiomyopathy, mitral regurgitation and various degrees of AV block had been described in the course of BMD[2]. There are few reports about complete atrioventricular block related with BMD and permanent cardiac pacemaker implantation as a treatment.

CASE REPORT

A 49-year-old man admitted to our emergency department with shortness of breath and fainting episodes for two weeks. He was suffering from muscle weakness for thirty years. He became unable to walk without help when he was 25 years old and became wheelchair bound ten years later. Becker Muscular Dystrophy was diagnosed in a university hospital when he was twenty years old but, he could not come to his control visits regularly.

In his family history, he has two sisters and two brothers.

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One of his brothers had died from a sudden death when he was 45 years old, we could not get any knowledge whether he had muscle disease or not. His other brother who was diagnosed as BMD and he has dilated cardiomyopathy and complete AVB for two years.

Cardiac examination showed a heart rate of 30 beats/min irregular rhythm, S3 gallop and apical 2/6 systolic murmur. Sytolic and diastolic arterial pressures were 90/60mmHg, respectively. There was bilateral rales on lung examination. Neurological examination showed atrophies proximal of upper and lower limbs and pseudohypertrophies in the popliteal muscles (Fig.1). Upper limb extremity muscle strength was evaluated as 3/5, lower limb extremity muscle strength was 2/5 at proximal and 3/5 at distal. Sensorial examinations were within normal ranges. Nerve conduction velocity studies showed normal findings and electromyogram has revealed myopathic degeneration. Creatine phosphokinase was elevated (870 IU/dl, Normal: 0-150 IU/L) and the other routine biochemical and haematological blood tests were all within normal ranges. The chest X- ray examination revealed an enlarged cardiac siluette. The electrocardiogram showed complete A-V block with varying heart rate between 30-36 beats/min (Fig.2). The echocardiogram showed marked increase in the size of left and right heart chambers, relatively thin walls and marked diminished contractility. Left ventricular EF was calculated as %30 with Teicholz method (figure 3). Cardiac valvular structures were all normal. There was mild to moderate degree of mitral regurgitation on Doppler investigation. It had been shown by genetic analysis that there was deletion of exons 45, 46 and 47 of the dystrophin gene of the patient's DNA, using the polymerase chain reaction and its extent was confirmed by Southern blotting (Fig. 4).

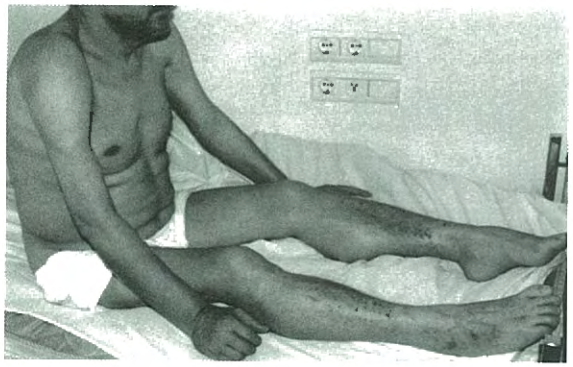

Fig. 1: Demonstrating the atrophies in the quadriceps muscles and pseudohypertrophies in the popliteal muscles.

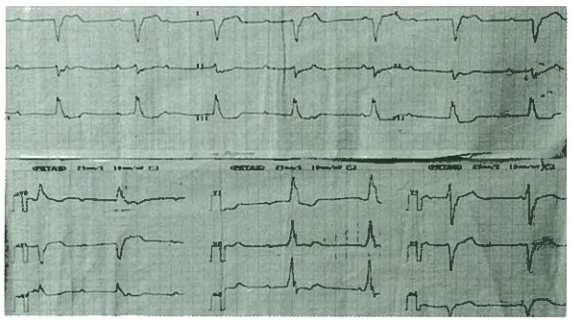

Fig. 2: ECG shows complete atrioventricular block .

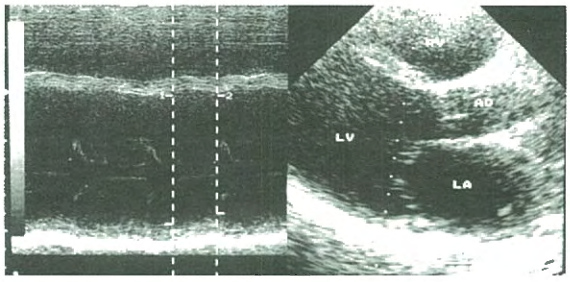

Fig. 3: Echocardiography shows severe left atrial and left ventricular dilatation and left ventricular systolic dysfunction. LV: Left Ventricle, LA: Left Atrium

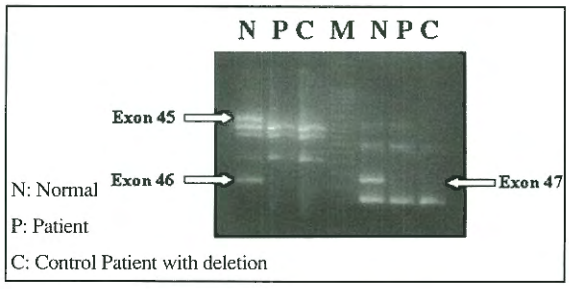

Fig. 4: Genetic analysis shows deletions of exons 45, 46 and 47 of the dystrophin gene of the patient's DNA, using the polymerase chain reaction

(N:Normal, P: Patient, C: Control patient with deletion)

As a result, dilated cardiomyopathy and complete AVB were diagnosed. A permanent cardiac pacemaker (DDDR) was implanted via right sub-clavian venous route. In addition, orally drugs for heart failure including digoxin, ACE inhibitor, furosemid and long acting nitrate was initiated as heart failure treatment. After implantation of permanent pacemaker and initiation of the heart failure treatment, fatigue and fainting episodes were disappeared and blood pressure reached up to an average of 120/80

mmHg and the heart rate of 70 bpm. There was not any complication following the procedure. After sixth months, the patient was haemodinamically stable and had any cardiac complaints.

DISCUSSION

There is abnormal dystrophin in the myocardium and skeletal muscle in BMD and DMD[3]. It might be expected that, severe myocardial involvement in patients with DMD who lack dystrophin in the myocardium and skeletal muscle compared to BMD[4]. Patients with DMD occasionally die from lung diseases. On the other hand, cardiac dysfunction is a major cause of death in patients with BMD and its mechanism is still unknown. Before the discovery of the dystrophin gene, the diagnosis of BMD and DMD was so difficult because of their clinical and pathological similarities with the other muscular dystrophies. Our patient was diagnosed by dystrophin analysis in addition to clinical and the other laboratory findings. In typical BMD, deletions almost always preserve reading frame of the gene and commonly involve exones 45, 47, 48 and 45 to 49 in the distal rod domain[5,6]. Our patient has genetic and clinical similarities with those findings. He has deletions of exones 45, 46 and 47 of the dytrophin gene.

Muntoni et al. reported X-linked dilated cardiomyopathy with a dystrophin gene deletion[7]. They proposed that the brain promotor of the dystrophin gene induced high levels of transcription in skeletal muscle but not in the heart, so patients lacking this promotor gene manifested cardiomyopathy despite the absence of muscle weakness.

Saito et al. found that prominent R wave in lead V1, suggesting the posterior wall damage and a decreased R wave amplitude or prominent Q wave in lead I, aVL and V6 suggesting lateral wall damage was most frequently present on ECG findings[8]. Electrocardiographic findings vary according to the underlying deletions in muscular dystrophies. Hassan et al. reported a family showing muscular dystrophy and atrioventricular block with an X-linked hereditary transmission. Among a known pedigree of 101 family members, 12 males were found to have skeletal muscle involvement and six needed pacemakers around age 30 years. Unlike the X-linked muscular dystrophies of Duchenne and of Becker, the predominant skeletal involvement was in humeral muscles, was usually very mild, and did not produce incapacitation. Cardiac involvement consisted of various atrial arrhythmias and atrioventricular block. They concluded that recognition of this subtle muscular dystrophy is important for early detection of incipient complete atrioventricular block to prevent fatal complications by pacemaker insertion [9]. Ducceschi et al. evaluated the arrhythmic profile in a population of 20 BMD patients searching for possible correlations between the severity of the arrhythmic events, the cardiac autonomic balance (assessed by heart rate variability analysis in the time domain) and the degree of left ventricular systolic impairment. A population of 14 male healthy individuals served as the control group. Finally they concluded that, in BMD there is cardiac autonomic imbalance characterized by sympathetic predominance and an increased susceptibility to ventricular arrhythmias, even in the absence of overt cardiomyopathy. Furthermore, the severity of the arrhythmic profile in BMD appears closely related to the degree of left ventricular systolic dysfunction[10].

Dilated cardiomyopathy is the most common form of cardiac damage of BMD[8]. So many deletions of exones of the dystrophin gene which could be associated with dilated cardiomyopathy had been described before. Saotome et al. reported a muscular dystrophy with exon-4 deletion[11]. Yu et al. reported a family with BMD presenting with cardiac involvement; the proband was a 41-year-old Japanese man who was hospitalized with exertional dyspnea and muscle weakness. Cardiac examination showed findings consistent with dilated cardiomyopathy. Dystrophin immunohistochemical analysis showed a discontinuous patchy staining pattern in cardiac and skeletal muscles biopsied from the proband. His brothers had high creatine kinase (CK) activity and abnormal electrocardiogram. Dystrophin gene analysis revealed that the proband and his brothers had G-to-T transversion at the terminal nucleotide of exon13. They concluded that the mutated dystrophin gene may cause cardiac involvement as a symptom precedent to skeletal muscle involvement[12]. Cardiac damages in our case were